



CREBBP gene

CREB binding protein

Normal Function

The *CREBBP* gene provides instructions for making CREB binding protein, which regulates the activity of many genes in tissues throughout the body. This protein plays an essential role in controlling cell growth and division and prompting cells to mature and assume specialized functions (differentiate). Studies in animals suggest that this protein may also be involved in the formation of long-term memories. CREB binding protein appears to be critical for normal development before and after birth.

CREB binding protein carries out its function by activating transcription, the process of making a blueprint of a gene for protein production. Specifically, CREB binding protein connects transcription factors, which are proteins that start the transcription process, with the complex of proteins that carries out transcription. On the basis of this function, CREB binding protein is called a transcriptional coactivator.

Health Conditions Related to Genetic Changes

Rubinstein-Taybi syndrome

A loss of one copy of the *CREBBP* gene in each cell causes Rubinstein-Taybi syndrome. In some cases, this loss occurs when a chromosomal rearrangement disrupts the region of chromosome 16 containing the gene. In other cases, mutations within the *CREBBP* gene itself are responsible for the condition. More than 90 mutations have been identified, including deletions and insertions of genetic material in the gene and changes in single DNA building blocks (nucleotides).

If one copy of the *CREBBP* gene is deleted or mutated, cells make only half of the normal amount of functional CREB binding protein. Although researchers are uncertain how a reduction in the amount of this protein leads to the specific features of Rubinstein-Taybi syndrome, it is clear that the loss of one copy of the *CREBBP* gene disrupts normal development before and after birth.

cancers

Genetic changes involving the *CREBBP* gene have been associated with certain types of cancer. These mutations are somatic, which means they are acquired during a person's lifetime and are present only in certain cells. In some cases, chromosomal rearrangements (translocations) disrupt the region of chromosome 16 that contains the *CREBBP* gene. For example, researchers have found a translocation between chromosome 8 and chromosome 16 in some people with a cancer of blood-

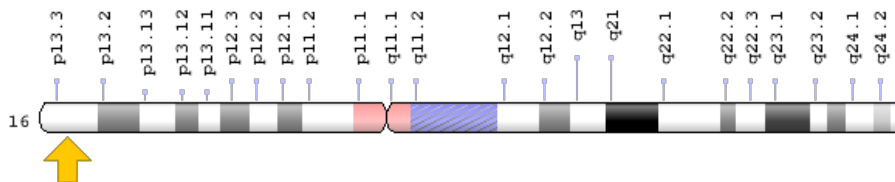
forming cells called acute myeloid leukemia (AML). Another translocation, involving chromosomes 11 and 16, has been found in some people who have undergone cancer treatment. This chromosomal change is associated with the later development of AML and two other cancers of blood-forming tissues (chronic myelogenous leukemia and myelodysplastic syndrome). These are sometimes described as treatment-related cancers because the translocation between chromosomes 11 and 16 occurs following chemotherapy for other forms of cancer.

Somatic mutations in the *CREBBP* gene also have been identified in several cases of ovarian cancer. Alterations in the *CREBBP* gene in ovarian cells lead to the production of an abnormally short, inactive version of CREB binding protein. A loss of this protein disrupts the normal regulation of cell growth and division, which can allow cancerous tumors to form. Researchers are working to determine the role of *CREBBP* mutations in the development and progression of ovarian cancers.

Chromosomal Location

Cytogenetic Location: 16p13.3, which is the short (p) arm of chromosome 16 at position 13.3

Molecular Location: base pairs 3,725,054 to 3,880,727 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CBP
- CBP_HUMAN
- CREB binding protein (Rubinstein-Taybi syndrome)

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): Chromosomal Abnormalities Involving Transcriptional Coactivators
<https://www.ncbi.nlm.nih.gov/books/NBK12465/#A44443>
- Molecular Biology of the Cell (fourth edition, 2002): Eucaryotic Gene Regulatory Proteins Often Assemble into Complexes on DNA
<https://www.ncbi.nlm.nih.gov/books/NBK26872/#A1300>

GeneReviews

- Rubinstein-Taybi Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1526>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CREBBP%5BTI%5D%29+OR+%28CREB+binding+protein%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- CREB-BINDING PROTEIN
<http://omim.org/entry/600140>
- LEUKEMIA, ACUTE MYELOID
<http://omim.org/entry/601626>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/CBPID42.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CREBBP%5Bgene%5D>
- HGNC Gene Family: Lysine acetyltransferases
<http://www.genenames.org/cgi-bin/genefamilies/set/486>
- HGNC Gene Family: Zinc fingers ZZ-type
<http://www.genenames.org/cgi-bin/genefamilies/set/91>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2348

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1387>
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<http://www.uniprot.org/uniprot/Q92793>

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